

Sheila L Unger

List of Publications by Year in descending order

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142
papers

6,904
citations

57631

44
h-index

69108

77
g-index

148
all docs

148
docs citations

148
times ranked

9104
citing authors

#	ARTICLE	IF	CITATIONS
1	Clinical and Molecular Diagnosis of Osteocraniostenosis in Fetuses and Newborns: Prenatal Ultrasound, Clinical, Radiological and Pathological Features. <i>Genes</i> , 2022, 13, 261.	1.0	5
2	Analysis of missense variants in the human genome reveals widespread gene-specific clustering and improves prediction of pathogenicity. <i>American Journal of Human Genetics</i> , 2022, 109, 457-470.	2.6	29
3	Cancer surveillance in children with Ollier Disease and Maffucci Syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 1338-1340.	0.7	2
4	Non-coding deletions identify Maenli lncRNA as a limb-specific En1 regulator. <i>Nature</i> , 2021, 592, 93-98.	13.7	53
5	Immune deficiency, autoimmune disease and intellectual disability: A pleiotropic disorder caused by biallelic variants in the <i>TPP2</i> gene. <i>Clinical Genetics</i> , 2021, 99, 780-788.	1.0	4
6	Update Swiss guideline for counselling and testing for predisposition to breast, ovarian, pancreatic and prostate cancer. <i>Swiss Medical Weekly</i> , 2021, 151, w30038.	0.8	4
7	Chondrodysplasia and growth failure in children after early hematopoietic stem cell transplantation for non-oncologic disorders. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 517-527.	0.7	3
8	CSGALNACT1 congenital disorder of glycosylation: A mild skeletal dysplasia with advanced bone age. <i>Human Mutation</i> , 2020, 41, 655-667.	1.1	15
9	Is height important for quality of life in children with skeletal dysplasias?. <i>European Journal of Medical Genetics</i> , 2020, 63, 103816.	0.7	11
10	Physicians communicating with women at genetic risk of breast and ovarian cancer: Are we in the middle of the ford between contradictory messages and unshared decision making?. <i>PLoS ONE</i> , 2020, 15, e0240054.	1.1	6
11	Ligand Binding to the Collagen VI Receptor Triggers a Talin-to-RhoA Switch that Regulates Receptor Endocytosis. <i>Developmental Cell</i> , 2020, 53, 418-430.e4.	3.1	12
12	Non-invasive prenatal testing leading to a maternal diagnosis of Charcot-Marie-Tooth neuropathy. <i>Journal of Human Genetics</i> , 2020, 65, 1035-1038.	1.1	4
13	Combined Lung and Liver Transplantation for Short Telomere Syndrome. <i>Liver Transplantation</i> , 2020, 26, 840-844.	1.3	10
14	The Connective Tissue Disorder Associated with Recessive Variants in the SLC39A13 Zinc Transporter Gene (Spondylo-Dysplastic Ehlers-Danlos Syndrome Type 3): Insights from Four Novel Patients and Follow-Up on Two Original Cases. <i>Genes</i> , 2020, 11, 420.	1.0	9
15	Osteogenesis imperfecta: towards an individualised interdisciplinary care strategy to improve physical activity and quality of life. <i>Swiss Medical Weekly</i> , 2020, 150, w20285.	0.8	4
16	The Liberfarb syndrome, a multisystem disorder affecting eye, ear, bone, and brain development, is caused by a founder pathogenic variant in the PISD gene. <i>Genetics in Medicine</i> , 2019, 21, 2734-2743.	1.1	33
17	Severe Peripheral Joint Laxity is a Distinctive Clinical Feature of Spondylodysplastic-Ehlers-Danlos Syndrome (EDS)-B4GALT7 and Spondylodysplastic-EDS-B3GALT6. <i>Genes</i> , 2019, 10, 799.	1.0	13
18	Nosology and classification of genetic skeletal disorders: 2019 revision. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 2393-2419.	0.7	431

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19	Does the clinical phenotype of mucopolysaccharidosis-III ^B differ from its I ^A counterpart?: supporting facts in a cohort of 18 patients. <i>Clinical Dysmorphology</i> , 2019, 28, 7-16.	0.1	10
20	Constitutional mismatch repair deficiency-associated brain tumors: report from the European C4CMMRD consortium. <i>Neuro-Oncology Advances</i> , 2019, 1, vdz033.	0.4	23
21	Lamin B receptor-related disorder is associated with a spectrum of skeletal dysplasia phenotypes. <i>Bone</i> , 2019, 120, 354-363.	1.4	11
22	Hypomorphic mutations of TRIP11 cause odontochondrodysplasia. <i>JCI Insight</i> , 2019, 4, .	2.3	30
23	Complex cranio-vertebral malformation: disruption sequence or iniencephaly?. <i>Clinical Dysmorphology</i> , 2018, 27, 105-108.	0.1	0
24	Confirmation of spondylo-epi-metaphyseal dysplasia with joint laxity, EXOC6B type. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 2934-2935.	0.7	5
25	Current Care and Investigational Therapies in Achondroplasia. <i>Current Osteoporosis Reports</i> , 2017, 15, 53-60.	1.5	50
26	CMG2/ANTXR2 regulates extracellular collagen VI which accumulates in hyaline fibromatosis syndrome. <i>Nature Communications</i> , 2017, 8, 15861.	5.8	56
27	Genetic disorders of bone – An historical perspective. <i>Bone</i> , 2017, 102, 1-4.	1.4	5
28	Small patella syndrome: New clinical and molecular insights into a consistent phenotype. <i>Clinical Genetics</i> , 2017, 92, 676-678.	1.0	10
29	Mutations in Fibronectin Cause a Subtype of Spondylometaphyseal Dysplasia with ‘Corner Fractures’. <i>American Journal of Human Genetics</i> , 2017, 101, 815-823.	2.6	37
30	Chondroitin Sulfate N-acetylgalactosaminyltransferase-1 (CSGalNAcT-1) Deficiency Results in a Mild Skeletal Dysplasia and Joint Laxity. <i>Human Mutation</i> , 2017, 38, 34-38.	1.1	22
31	The Bone in Genetic and Metabolic Diseases: A Practical Approach. , 2017, , 371-380.		0
32	Loss-of-function mutations in the X-linked biglycan gene cause a severe syndromic form of thoracic aortic aneurysms and dissections. <i>Genetics in Medicine</i> , 2017, 19, 386-395.	1.1	94
33	Corner fracture type spondylometaphyseal dysplasia: Overlap with type II collagenopathies. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 733-739.	0.7	8
34	Sojourner in a foreign land. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 2594-2595.	0.7	0
35	NANS-mediated synthesis of sialic acid is required for brain and skeletal development. <i>Nature Genetics</i> , 2016, 48, 777-784.	9.4	125
36	Natural history and life-threatening complications in Myhre syndrome and review of the literature. <i>European Journal of Pediatrics</i> , 2016, 175, 1307-1315.	1.3	15

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37	Cortical-Bone Fragility – Insights from sFRP4 Deficiency in Pyle’s Disease. <i>New England Journal of Medicine</i> , 2016, 374, 2553-2562.	13.9	119
38	Novel de novo mutations in <i>ZBTB20</i> in Primrose syndrome with congenital hypothyroidism. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 1626-1629.	0.7	27
39	Brief Report: Peripheral Osteolysis in Adults Linked to <i>ASAHL1</i> (Acid Ceramidase) Mutations: A New Presentation of Farber’s Disease. <i>Arthritis and Rheumatology</i> , 2016, 68, 2323-2327.	2.9	17
40	Mutations in <i>LONP1</i> , a mitochondrial matrix protease, cause CODAS syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 1501-1509.	0.7	61
41	Mutations in the heat-shock protein A9 (HSPA9) gene cause the EVEN-PLUS syndrome of congenital malformations and skeletal dysplasia. <i>Scientific Reports</i> , 2015, 5, 17154.	1.6	65
42	NBAS mutations cause a multisystem disorder involving bone, connective tissue, liver, immune system, and retina. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 2902-2912.	0.7	66
43	Nosology and classification of genetic skeletal disorders: 2015 revision. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 2869-2892.	0.7	453
44	Leri’s pleonosteosis, a congenital rheumatic disease, results from microduplication at 8q22.1 encompassing <i>GDF6</i> and <i>SDC2</i> and provides insight into systemic sclerosis pathogenesis. <i>Annals of the Rheumatic Diseases</i> , 2015, 74, 1249-1256.	0.5	22
45	Challenges in managing genetic cancer risk: a long-term qualitative study of unaffected women carrying BRCA1/BRCA2 mutations. <i>Genetics in Medicine</i> , 2015, 17, 726-732.	1.1	17
46	Further delineation of the KAT6B molecular and phenotypic spectrum. <i>European Journal of Human Genetics</i> , 2015, 23, 1165-1170.	1.4	56
47	Stuve-Wiedemann syndrome with a novel mutation. <i>BMJ Case Reports</i> , 2015, 2015, bcr2015212032.	0.2	9
48	Positive effects of an angiotensin II type 1 receptor antagonist in Camurati’s Engelmann disease: A single case observation. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 2667-2671.	0.7	21
49	Eight years experience from a skeletal dysplasia referral center in a tertiary hospital in Southern India: A model for the diagnosis and treatment of rare diseases in a developing country. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 2317-2323.	0.7	18
50	Cono-spondylar dysplasia: Clinical, radiographic, and molecular findings of a previously unreported disorder. , 2014, 164, 2147-2152.		0
51	Acampomelic Form of Campomelic Dysplasia with SOX9 Missense Mutation. <i>Indian Journal of Pediatrics</i> , 2014, 81, 98-100.	0.3	8
52	<i>MMP13</i> mutations are the cause of recessive metaphyseal dysplasia, Spahr type. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 1175-1179.	0.7	14
53	CDK10/cyclin M is a protein kinase that controls ETS2 degradation and is deficient in STAR syndrome. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2013, 110, 19525-19530.	3.3	73
54	Chondrodysplasias. , 2013, , 1-45.		2

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55	Focal dermal hypoplasia (goltzâ€gorlin syndrome): A new case with a novel variant in the <i>PORCN</i> gene (c.1250T>C:p.F417S) and unusual spinal anomaly. American Journal of Medical Genetics, Part A, 2013, 161, 1750-1754.	0.7	5
56	Mutations in B3GALT6, which Encodes a Glycosaminoglycan Linker Region Enzyme, Cause a Spectrum of Skeletal and Connective Tissue Disorders. American Journal of Human Genetics, 2013, 92, 927-934.	2.6	112
57	FAM111A Mutations Result in Hypoparathyroidism and Impaired Skeletal Development. American Journal of Human Genetics, 2013, 92, 990-995.	2.6	114
58	Whole-Genome Analysis Reveals that Mutations in Inositol Polyphosphate Phosphatase-like 1 Cause Opsismodysplasia. American Journal of Human Genetics, 2013, 92, 137-143.	2.6	53
59	Comprehensive Clinical and Molecular Analysis of 12 Families with Type 1 Recessive Cutis Laxa. Human Mutation, 2013, 34, 111-121.	1.1	67
60	Homozygosity for a novel truncating mutation confirms <i>TBX15</i> deficiency as the cause of Cousin syndrome. American Journal of Medical Genetics, Part A, 2013, 161, 3161-3165.	0.7	14
61	Dyssegmental dysplasia, Silvermanâ€Handmaker type: prenatal ultrasound findings and molecular analysis. Prenatal Diagnosis, 2013, 33, 1039-1043.	1.1	7
62	Clinical and radiological findings in Pallisterâ€Killian syndrome. European Journal of Medical Genetics, 2012, 55, 167-172.	0.7	18
63	A Diagnostic Approach to Skeletal Dysplasias. , 2012, , 403-437.		6
64	Extracellular matrix and platelet function in patients with musculocontractural Ehlersâ€Danlos syndrome caused by mutations in the <i>CHST14</i> gene. American Journal of Medical Genetics, Part A, 2012, 158A, 1344-1354.	0.7	32
65	The diagnostic challenge of progressive pseudorheumatoid dysplasia (PPRD): A review of clinical features, radiographic features, and <i>WISP3</i> mutations in 63 affected individuals. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2012, 160C, 217-229.	0.7	74
66	TRPV4â€associated skeletal dysplasias. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2012, 160C, 190-204.	0.7	71
67	New topics in the skeletal dysplasias. , 2012, 160C, 143-144.		1
68	Recurrent Dominant Mutations Affecting Two Adjacent Residues in the Motor Domain of the Monomeric Kinesin KIF22 Result in Skeletal Dysplasia and Joint Laxity. American Journal of Human Genetics, 2012, 90, 170.	2.6	0
69	Pseudoachondroplasia and multiple epiphyseal dysplasia: A 7â€year comprehensive analysis of the known disease genes identify novel and recurrent mutations and provides an accurate assessment of their relative contribution. Human Mutation, 2012, 33, 144-157.	1.1	104
70	Molecular screening of ADAMTSL2 gene in 33 patients reveals the genetic heterogeneity of geleophysic dysplasia. Journal of Medical Genetics, 2011, 48, 417-421.	1.5	45
71	Genetic deficiency of tartrate-resistant acid phosphatase associated with skeletal dysplasia, cerebral calcifications and autoimmunity. Nature Genetics, 2011, 43, 132-137.	9.4	151
72	Chondrodysplasia and Abnormal Joint Development Associated with Mutations in IMPAD1, Encoding the Golgi-Resident Nucleotide Phosphatase, gPAPP. American Journal of Human Genetics, 2011, 88, 608-615.	2.6	88

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73	Mutations in the TGF β 2 Binding-Protein-Like Domain 5 of FBN1 Are Responsible for Acromioclavicular and Geleophysic Dysplasias. <i>American Journal of Human Genetics</i> , 2011, 89, 7-14.	2.6	199
74	Recurrent Dominant Mutations Affecting Two Adjacent Residues in the Motor Domain of the Monomeric Kinesin KIF22 Result in Skeletal Dysplasia and Joint Laxity. <i>American Journal of Human Genetics</i> , 2011, 89, 767-772.	2.6	31
75	Identification of signal peptide domain SOST mutations in autosomal dominant craniodiaphyseal dysplasia. <i>Human Genetics</i> , 2011, 129, 497-502.	1.8	68
76	Ala \rightarrow Asp \rightarrow Raas \rightarrow Rothschild (limb/pelvis/uterus \rightarrow hypoplasia/aplasia) syndrome and <i>WNT7A</i> mutations: Genetic homogeneity and nosological delineation. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 332-336.	0.7	19
77	Nosology and classification of genetic skeletal disorders: 2010 revision. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 943-968.	0.7	573
78	Axial spondylometaphyseal dysplasia: Additional reports. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 2521-2528.	0.7	8
79	Revisit of multiple epiphyseal dysplasia: Ethnic difference in genotypes and comparison of radiographic features linked to the <i>COMP</i> and <i>MATN3</i> genes. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 2669-2680.	0.7	20
80	Fetal akinesia in metatropic dysplasia: The combined phenotype of chondrodysplasia and neuropathy?. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 2860-2864.	0.7	30
81	Whole \rightarrow exome sequencing detects somatic mutations of <i>IDH1</i> in metaphyseal chondromatosis with <i>D</i> -hydroxyglutaric aciduria (MC \rightarrow HGA). <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 2609-2616.	0.7	47
82	A family with a new <i>elastin</i> gene mutation: broad clinical spectrum, including sudden cardiac death. <i>Cardiology in the Young</i> , 2011, 21, 62-65.	0.4	18
83	<i>CANT1</i> mutation is also responsible for Desbuquois dysplasia, type 2 and Kim variant. <i>Journal of Medical Genetics</i> , 2011, 48, 32-37.	1.5	39
84	A variant of Desbuquois dysplasia characterized by advanced carpal bone age, short metacarpals, and elongated phalanges: Report of seven cases. <i>American Journal of Medical Genetics, Part A</i> , 2010, 152A, 875-885.	0.7	32
85	Spondylo \rightarrow epiphyseal dysplasia, Maroteaux type (pseudo \rightarrow Marquo syndrome type 2), and parastremmatic dysplasia are caused by <i>TRPV4</i> mutations. <i>American Journal of Medical Genetics, Part A</i> , 2010, 152A, 1443-1449.	0.7	56
86	Phenotypic features of carbohydrate sulfotransferase 3 (<i>CHST3</i>) deficiency in 24 patients: Congenital dislocations and vertebral changes as principal diagnostic features. <i>American Journal of Medical Genetics, Part A</i> , 2010, 152A, 2543-2549.	0.7	67
87	A familial case of tricho \rightarrow rhino \rightarrow phalangeal syndrome type III with a novel missense mutation in exon 6 of the <i>TRPS1</i> gene. <i>Journal of the European Academy of Dermatology and Venereology</i> , 2010, 24, 612-614.	1.3	2
88	St \rightarrow ve \rightarrow Wiedemann syndrome: long \rightarrow term follow \rightarrow up and genetic heterogeneity. <i>Clinical Genetics</i> , 2010, 77, 266-272.	1.0	33
89	<i>TRPV4</i> -pathy, a novel channelopathy affecting diverse systems. <i>Journal of Human Genetics</i> , 2010, 55, 400-402.	1.1	45
90	Novel and recurrent <i>TRPV4</i> mutations and their association with distinct phenotypes within the <i>TRPV4</i> dysplasia family. <i>Journal of Medical Genetics</i> , 2010, 47, 704-709.	1.5	58

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91	Phenotypic and molecular characterization of a novel case of dyssegmental dysplasia, Silverman-Handmaker type. <i>European Journal of Medical Genetics</i> , 2010, 53, 294-298.	0.7	10
92	Identification of loss-of-function mutations of SLC35D1 in patients with Schneckenbecken dysplasia, but not with other severe spondylodysplastic dysplasias group diseases. <i>Journal of Medical Genetics</i> , 2009, 46, 562-568.	1.5	41
93	A distinct form of spondyloepimetaphyseal dysplasia with joint laxity (SEMDIL)-leptodactylic type: radiological characteristics in seven new patients. <i>Skeletal Radiology</i> , 2009, 38, 803-811.	1.2	9
94	A large-scale mutation search reveals genetic heterogeneity in 3M syndrome. <i>European Journal of Human Genetics</i> , 2009, 17, 395-400.	1.4	48
95	Refinement of the 12q14 microdeletion syndrome: primordial dwarfism and developmental delay with or without osteopoikilosis. <i>European Journal of Human Genetics</i> , 2009, 17, 1141-1147.	1.4	33
96	Mutations in the Heparan-Sulfate Proteoglycan Glypican 6 (GPC6) Impair Endochondral Ossification and Cause Recessive Omodysplasia. <i>American Journal of Human Genetics</i> , 2009, 84, 760-770.	2.6	106
97	Mutations in MMP9 and MMP13 Determine the Mode of Inheritance and the Clinical Spectrum of Metaphyseal Anadysplasia. <i>American Journal of Human Genetics</i> , 2009, 85, 168-178.	2.6	54
98	Mutations in MMP9 and MMP13 Determine the Mode of Inheritance and the Clinical Spectrum of Metaphyseal Anadysplasia. <i>American Journal of Human Genetics</i> , 2009, 85, 420.	2.6	0
99	Fetal MR imaging of atelosteogenesis type II (AO-II). <i>Pediatric Radiology</i> , 2008, 38, 1345-1349.	1.1	14
100	Clinical and radiographic delineation of odontochondrodysplasia. <i>American Journal of Medical Genetics, Part A</i> , 2008, 146A, 770-778.	0.7	23
101	Chondrodysplasia punctata associated with maternal autoimmune diseases: Expanding the spectrum from systemic lupus erythematosus (SLE) to mixed connective tissue disease (MCTD) and scleroderma report of eight cases. <i>American Journal of Medical Genetics, Part A</i> , 2008, 146A, 3038-3053.	0.7	38
102	Congenital Joint Dislocations Caused by Carbohydrate Sulfotransferase 3 Deficiency in Recessive Larsen Syndrome and Humero-Spinal Dysostosis. <i>American Journal of Human Genetics</i> , 2008, 82, 1368-1374.	2.6	92
103	Congenital Joint Dislocations Caused by Carbohydrate Sulfotransferase 3 Deficiency in Recessive Larsen Syndrome and Humero-Spinal Dysostosis. <i>American Journal of Human Genetics</i> , 2008, 83, 293.	2.6	0
104	TBX15 Mutations Cause Craniofacial Dysmorphism, Hypoplasia of Scapula and Pelvis, and Short Stature in Cousin Syndrome. <i>American Journal of Human Genetics</i> , 2008, 83, 649-655.	2.6	60
105	Mutations in the cyclin family member FAM58A cause an X-linked dominant disorder characterized by syndactyly, telecanthus and anogenital and renal malformations. <i>Nature Genetics</i> , 2008, 40, 287-289.	9.4	45
106	Multiple epiphyseal dysplasia: clinical and radiographic features, differential diagnosis and molecular basis. <i>Best Practice and Research in Clinical Rheumatology</i> , 2008, 22, 19-32.	1.4	56
107	Defective C-propeptides of the Pro α 2(I) Chain of Type I Procollagen Impede Molecular Assembly and Result in Osteogenesis Imperfecta. <i>Journal of Biological Chemistry</i> , 2008, 283, 16061-16067.	1.6	57
108	The Zinc Transporter SLC39A13/ZIP13 Is Required for Connective Tissue Development; Its Involvement in BMP/TGF- β 2 Signaling Pathways. <i>PLoS ONE</i> , 2008, 3, e3642.	1.1	240

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109	The Shwachman-Bodian-Diamond syndrome gene mutations cause a neonatal form of spondylometaphyseal dysplasia (SMD) resembling SMD Sedaghatian type. <i>Journal of Medical Genetics</i> , 2007, 44, e73-e73.	1.5	14
110	Nosology and classification of genetic skeletal disorders: 2006 revision. <i>American Journal of Medical Genetics, Part A</i> , 2007, 143A, 1-18.	0.7	301
111	COL2A1-related skeletal dysplasias with predominant metaphyseal involvement. <i>American Journal of Medical Genetics, Part A</i> , 2007, 143A, 161-167.	0.7	32
112	<i>Filamin A</i> mutation is one cause of FG syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2007, 143A, 1876-1879.	0.7	44
113	œœs NF1 a genetic skeletal disorder?œœ”A response. <i>American Journal of Medical Genetics, Part A</i> , 2007, 143A, 2084-2084.	0.7	10
114	Raine syndrome: A rare lethal osteosclerotic bone dysplasia. Prenatal diagnosis, autopsy, and neuropathological findings. <i>American Journal of Medical Genetics, Part A</i> , 2007, 143A, 3280-3285.	0.7	26
115	Preselection of cases through expert clinical and radiological review significantly increases mutation detection rate in multiple epiphyseal dysplasia. <i>European Journal of Human Genetics</i> , 2007, 15, 150-154.	1.4	28
116	A molecular and clinical study of Larsen syndrome caused by mutations in FLNB. <i>Journal of Medical Genetics</i> , 2006, 44, 89-98.	1.5	102
117	X-Linked dominant chondrodysplasia punctata: prenatal diagnosis and autopsy findings. <i>Prenatal Diagnosis</i> , 2006, 26, 1235-1240.	1.1	10
118	Petrified ears in a patient with Keutel syndrome: temporal bone CT findings. <i>Pediatric Radiology</i> , 2006, 36, 241-243.	1.1	13
119	Spondyloenchondrodysplasia with spasticity, cerebral calcifications, and immune dysregulation: Clinical and radiographic delineation of a pleiotropic disorder. <i>American Journal of Medical Genetics, Part A</i> , 2006, 140A, 541-550.	0.7	58
120	Fetus with two identical reciprocal translocations: Description of a rare complication of consanguinity. <i>American Journal of Medical Genetics, Part A</i> , 2006, 140A, 769-774.	0.7	12
121	Contiguous hemizygous deletion of TBX5, TBX3, and RBM19 resulting in a combined phenotype of Holt-Oram and ulnar-mammary syndromes. <i>American Journal of Medical Genetics, Part A</i> , 2006, 140A, 1880-1886.	0.7	29
122	Immunohistochemical Evaluation of Conjunctival Fibrillin-1 in Marfan Syndrome. <i>JAMA Ophthalmology</i> , 2006, 124, 205.	2.6	17
123	The mildest form of campomelic dysplasia. <i>American Journal of Medical Genetics, Part A</i> , 2005, 132A, 113-113.	0.7	9
124	Mucopolipidosis II presenting as severe neonatal hyperparathyroidism. <i>European Journal of Pediatrics</i> , 2005, 164, 236-243.	1.3	37
125	Evolutionary Comparison Provides Evidence for Pathogenicity of RMRP Mutations. <i>PLoS Genetics</i> , 2005, 1, e47.	1.5	57
126	IGF-I Receptor Mutations and Intrauterine and Postnatal Growth Retardation. <i>New England Journal of Medicine</i> , 2004, 350, 1362-1363.	13.9	1

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127	Null Leukemia Inhibitory Factor Receptor (LIFR) Mutations in StÅ½ve-Wiedemann/Schwartz-Jampel Type 2 Syndrome. American Journal of Human Genetics, 2004, 74, 298-305.	2.6	162
128	Mental Retardation and Abnormal Skeletal Development (Dyggve-Melchior-Clausen Dysplasia) Due to Mutations in a Novel, Evolutionarily Conserved Gene. American Journal of Human Genetics, 2003, 72, 419-428.	2.6	53
129	A Diagnostic Approach to Skeletal Dysplasias. , 2003, , 375-IX.		7
130	A Genetic Approach to the Diagnosis of Skeletal Dysplasia. Clinical Orthopaedics and Related Research, 2002, 401, 32-38.	0.7	30
131	Evidence That Smith-McCort Dysplasia and Dyggve-Melchior-Clausen Dysplasia Are Allelic Disorders That Result from Mutations in a Gene on Chromosome 18q12. American Journal of Human Genetics, 2002, 71, 947-951.	2.6	23
132	Severe cleidocranial dysplasia can mimic hypophosphatasia. European Journal of Pediatrics, 2002, 161, 623-626.	1.3	46
133	"Duplicate calcaneus": a rare developmental defect observed in several skeletal dysplasias. Pediatric Radiology, 2001, 31, 38-42.	1.1	17
134	Multiple epiphyseal dysplasia: radiographic abnormalities correlated with genotype. Pediatric Radiology, 2001, 31, 10-18.	1.1	61
135	New dysplasia or achondrogenesis type 1B? The importance of histology and molecular biology in delineating skeletal dysplasias. Pediatric Radiology, 2001, 31, 893-894.	1.1	7
136	Double heterozygosity for pseudoachondroplasia and spondyloepiphyseal dysplasia congenita. American Journal of Medical Genetics Part A, 2001, 104, 140-146.	2.4	42
137	Pseudoachondroplasia and multiple epiphyseal dysplasia: New etiologic developments. American Journal of Medical Genetics Part A, 2001, 106, 244-250.	2.4	125
138	Oto-palato-digital syndrome, type II: Report of three cases with further delineation of the chondro-osseous morphology. American Journal of Medical Genetics Part A, 2000, 95, 193-200.	2.4	16
139	Early-infantile galactosialidosis: Prenatal presentation and postnatal follow-up. American Journal of Medical Genetics Part A, 1999, 85, 38-47.	2.4	44
140	Partial tetrasomy with triplication of chromosome (5) (p14-p15.33) in a patient with severe multiple congenital anomalies. American Journal of Medical Genetics Part A, 1998, 79, 103-107.	2.4	32
141	Partial tetrasomy with triplication of chromosome (5) (p14-p15.33) in a patient with severe multiple congenital anomalies. , 1998, 79, 103.		2
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